Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Occlusion of the Bowel and Peritoneal Carcinosis Revealing Peutz-Jeghers Syndrome: A Case Report

S. Outaghyame[1*], H. Loukili[1], Y. Bouktib[1], A. El Hajjami[1], B. Boutakioute[1], M. Ouali Idrissi[1], N. Cherif Idrissi[1]

[1]Radiology Department, Er-Razi Hospital, CHU Mohamed VI Marrakech, Morocco

DOI: 10.36347/sjmcr.2024.v12i03.026 | **Received:** 02.02.2023 | **Accepted:** 11.03.2024 | **Published:** 29.03.2024

***Corresponding author:** S. Outaghyame
Radiology Department, Er-Razi Hospital, CHU Mohamed VI Marrakech, Morocco

Abstract **Case Report**

Peutz-Jeghers syndrome (PJS) is characterised by the association of hamartomatous digestive polyposis and mucocutaneous lentiginosis. Patients are at risk of mechanical and haemorrhagic complications. It is a syndrome predisposing to cancer. We report the case of a 44-year-old man with a history of brown pigment stains around the mouth at the age of 10, which disappeared in adulthood, followed by bowel resections at 3, 16 and 18 years of age for occlusive episodes, having presented a month ago with diffuse abdominal distension complicated within three days by an occlusive syndrome with abdominal pain of moderate intensity, with no other associated digestive signs including digestive or extra digestive haemorrhage, all evolving in a context of altered general condition with weight loss. On clinical examination, the flanks were dull, there was diffuse abdominal tenderness and a median laparotomy scar. The patient underwent FOGD and colonoscopy, which revealed a mucosa lined with gastro-duodenal and colonic polyps of varying size and suspicious appearance. A biopsy of these polypoid formations was performed, confirming Peutz-Jeghers syndrome. A cytological and immunohistochemical study of the ascites fluid found atypical carcinomatous cells. In the literature, Peutz-Jeghers syndrome may be revealed clinically or at the stage of complications such as haemorrhage, intussusception or intestinal obstruction. As in the case of our patient, who presented with an occlusive syndrome with no detectable digestive haemorrhage. Peutz-Jeghers syndrome is a rare condition. However, it is important for clinicians to be aware of it and to consider its possibility in cases of occlusive syndrome with peri-orificial lentiginosis in young adults.

Keywords: Peutz-Jeghers, Polyps, Bowel Obstruction, Peritoneal Carcinosis, CT scan.

INTRODUCTION

Peutz-Jeghers syndrome (PJS) is characterised by a combination of digestive polyposis and mucocutaneous lentiginosis, predominantly periorificial. It is a rare, hereditary, autosomal dominant condition. It is sometimes associated with malignant digestive or extra-digestive tumours. Intestinal polyposis is the essential component [1]. We report the case of a 44-year-old man who presented with an occlusive syndrome over intestinal polyposis, revealing Peutz-Jeghers syndrome with signs of peritoneal carcinosis.

The aim of this study is to gain an understanding of this syndrome, its main complications and therapeutic advances in the management of this syndrome in the light of a review of the literature. We will also highlight the importance of periodic monitoring to detect the occurrence of cancer.

OBSERVATION

This is a 44-year-old man with a history of brown pigment stains around the mouth at the age of 10, which disappeared over time into adulthood, then notion of bowel resections at 3, 16 and 18 years for occlusive episodes, consulted emergency for diffuse abdominal distension evolving for a month, This symptomatology worsened 3 days ago with the appearance of diffuse abdominal pain of moderate intensity, with no triggering factor, no connection with eating and no analgesic position, and no other associated digestive signs, in particular no vomiting or digestive haemorrhage, all evolving in a context of altered general condition with weight loss.

On clinical examination, the flanks were dull and sloping, there was diffuse abdominal tenderness with conjunctival sub-icterus, and oedema of the lower limbs with abdominal scarring; this was evidence of a median laparotomy.

The patient initially underwent FOGD and colonoscopy, which revealed a mucosa lined with gastro-duodenal and colonic polyps of varying size and suspicious appearance. A biopsy of these polypoid formations was performed, confirming Peutz-Jeghers syndrome, even in the absence of labial lentiginosis.

A cytological and immunohistochemical study of the ascites fluid was then carried out, revealing a protein-rich fluid with the presence of atypical carcinomatous cells.

An abdominal ultrasound was performed, revealing a very large peritoneal effusion, anechoic, partitioned in places, with no detectable digestive thickening.

An additional abdominal CT scan (Figure 1) showed multiple endoluminal polyps involving the greco-colonic anes, one of which was obstructive at the jujenal level.

Associated with this was a very large, compartmentalised peritoneal effusion associated with peritoneal implants and significant diffuse infiltration of the mesenteric and epiploic fat, and thickening of the peritoneal layers, all suggestive of peritoneal carcinosis. A macrolitis gallbladder was reported. (Figure 1)

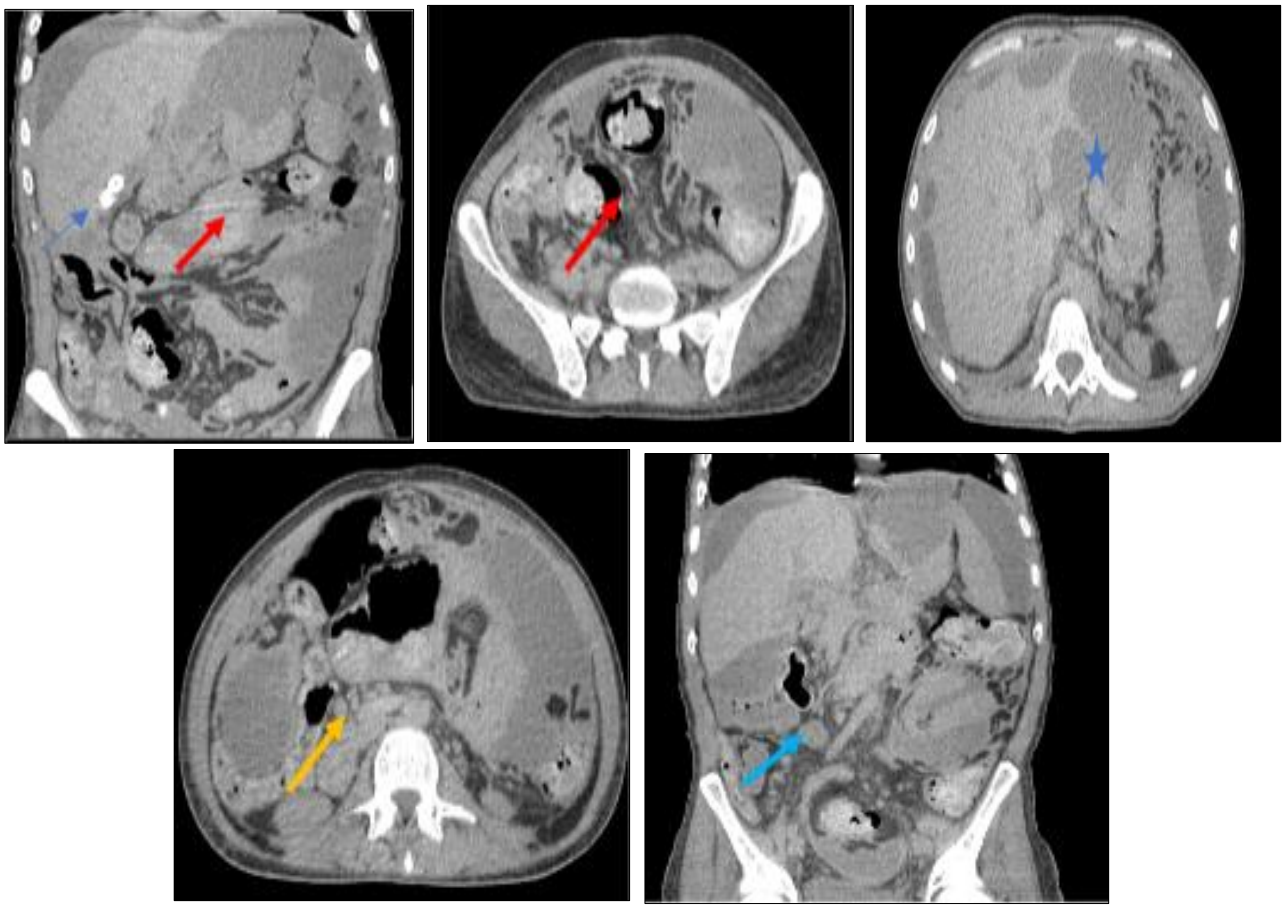

Figure 1: Abdominal CT scan with PDC injection objective:

- Multiple endoluminal polyps involving the grelo-colonic ansae (red arrow), within the framework of Peutz-Jeghers syndrome (PJS), one of which is obstructive at the jujenal level. (Orange arrow)
- Associated peritoneal effusion of great abundance (star) associated with scattered peritoneal implants, abdominal adenopathy (blue arrow) and extensive diffuse infiltration of mesenteric and epiploic fat, producing the epiploic cake sign, with thickening of the peritoneal sheets, in connection with peritoneal carcinosis.
- Macrolitic gallbladder. (Thin arrow)

DISCUSSION

Clinical criteria for a definite diagnosis of PJS include the presence of a hamartoma associated with two of the following three signs: mucocutaneous pigmentation, small bowel polyposis or a family history of PJS [1-3]. Pigmentation usually appears in the first year of life, but in adulthood it may fade and in some cases even disappear [4]. Pigmentation of the lips and oral mucosa is a key feature, playing an important role in early diagnosis. In this patient's case, the pigmentation consisted of spots on the lips and hamartomatous polyps in the gastrointestinal tract. However, some patients have only mucocutaneous pigmentation, which is defined as incomplete SJP [5].

The polyps found in SJP generally appear in adolescence and early adulthood. They develop in the first decade of life and most patients become symptomatic between the ages of 10 and 30 [3, 4]. Patients usually present in the first decade with polyp-related complications such as abdominal pain, intestinal obstruction, intussusception and overt or occult gastrointestinal bleeding [6]. Our patient presented with symptoms of upper bowel obstruction and copious peritoneal effusion.

The etiology of PJS is unclear. Mutations of the STK11 gene seems to be responsible for PJS, while STK 11 is a tumor suppressor gene and encodes a serine/threonine kinase and maps to chromosome 19p13.3 [7, 8]. STK11 play a role of tumor suppressor in cells. Loss of STK11 protein kinase activity is associated with occurrence and development of tumor. So patients with PJS have an increasing incidence of malignant tumors compared with the general population. The most common malignancy associated with PJS is colorectal cancer, followed by breast, small bowel, gastric, and pancreatic cancers, with lifetime cumulative cancer risks up to 93% [5-9].

In Peutz-Jeghers syndrome, malignant tumors are considered to appear in adulthood, and rarely occur in children, this was the case in our patient who presented with signs of peritoneal carcinosis.[10] . Patients should be instructed on the need for cancer surveillance, each patient is scheduled for follow-up abdominal ultrasound examination (liver gallbladder, pancreas, spleen) every six months and upper gastrointestinal endoscopy every three years according to the guidelines for PJS. Combined endoscopic and surgical treatment has been reported and is considered to be the best in terms of quality of life [11, 12]. Surgical resection may appropriately be used for the patients with giant or complication of polyp such as obstruction, intussusceptions or gastrointestinal bleeding.

Recently, some experts have used double balloon enteroscopy removal small bowel PJS polyps, which may decrease the need for laparotomy.

CONCLUSION

Peutz-Jeghers syndrome is a rare condition. It is a syndrome with a predisposition to cancer. Our study showed hamartomatous Peutz-Jeghers polyps complicated by several occlusive episodes progressing to peritoneal carcinosis. Endoscopic polypectomy is of great value in the management of this syndrome, as it avoids the need for repeated surgery. A monitoring protocol must be established for each PJS patient throughout their life to avoid complications. Although the frequency of this syndrome is low, it is important for clinicians to be aware of it and to consider its possibility in cases of digestive haemorrhage or occlusive syndrome.

BIBLIOGRAPHIE

1. Arber, N., & Moshkowitz, M. (2011). Small bowel polyposis syndromes. *Current gastroenterology reports*, *13*, 435-441.
2. Calva, D., & Howe, J. R. (2008). Hamartomatous polyposis syndromes. *Surgical Clinics of North America*, *88*(4), 779-817.
3. Giardiello, F. M., & Trimbath, J. D. (2006). Peutz-Jeghers syndrome and management recommendations. *Clinical Gastroenterology and Hepatology*, *4*(4), 408-415.
4. Szanto, P., Barbieru, V., Badea, R., Pop, T., Rusu, I., & Al Hajjar, N. (2014). Unexpected Peutz-Jeghers syndrome in an adult presenting with intermittent upper intestinal obstruction. A case report. *J Gastrointestin Liver Dis*, *23*(1), 91-94.
5. Fan, R. Y., & Sheng, J. Q. (2015). Un cas de syndrome de Peutz-Jeghers associé à une néoplasie intramuqueuse de haut grade. *Int J Clin Exp Pathol*, *8*, 7503-5.
6. Kopacova, M., Tacheci, I., & Rejchrt, S. (2009). Peutz-Jeghers syndrome: diagnostic et approche thérapeutique. *World J Gastroenterol*, *15*, 5397-408.
7. Hemminki, A., Markie, D., Tomlinson, I., Avizienyte, E., Roth, S., Loukola, A., ... & Aaltonen, L. A. (1998). A serine/threonine kinase gene defective in Peutz–Jeghers syndrome. *Nature*, *391*(6663), 184-187.
8. Huang, Z., Miao, S., Wang, L., Zhang, P., Wu, B., Wu, J., & Huang, Y. (2015). Clinical characteristics and STK11 gene mutations in Chinese children with Peutz-Jeghers syndrome. *BMC gastroenterology*, *15*, 1-8.
9. Van Lier, M. G. F., Wagner, A., Mathus-Vliegen, E. M. H., Kuipers, E. J., Steyerberg, E. W., & van Leerdam, M. E. (2010). High cancer risk in Peutz–Jeghers syndrome: a systematic review and surveillance recommendations. *Official journal of the American College of Gastroenterology| ACG*, *105*(6), 1258-1264.
10. Wangler, M. F., Chavan, R., Hicks, M. J., Nuchtern, J. G., Hegde, M., Plon, S. E., & Thompson, P. A. (2013). Unusually early presentation of small-bowel adenocarcinoma in a patient with Peutz-Jeghers syndrome. *Journal of pediatric hematology/oncology*, *35*(4), 323-328.
11. Lin, B. C., Lien, J. M., Chen, R. J., Fang, J. F., & Wong, Y. C. (2000). Combined endoscopic and surgical treatment for the polyposis of Peutz-Jeghers syndrome. *Surgical endoscopy*, *14*(12), 1185-1187.
12. Seenath, M. M., Scott, M. J., Morris, A. I., Ellis, A., & Hershman, M. J. (2003). Combined surgical and endoscopic clearance of small-bowel polyps in Peutz-Jeghers syndrome. *Journal of the Royal Society of Medicine*, *96*(10), 505-506.